

SUMMARY

Lipoid proteinosis is a rare recessively inherited multisystem disorder which primarily affects the skin, oral cavity and larynx.

The characteristic abnormality of this disease is the deposition of an amorphous hyaline material in the skin and mucous membranes.

The hyaline material which is strongly periodic acid Schiff positive, diastase resistant is present in thick homogeneous bands in the upper dermis and in patchy distribution in the lower dermis about blood vessels, sweat glands and dermal nerves.

Early investigators suspected that the disorder is a lipoidosis and the hyaline is at least partially or completely derived from blood plasma. Other investigators suggested the origin to be degenerated fibers of collagen and elastic tissues.

Recently it was reported that the disease is a primary disturbance of collagen metabolism and the material is principally glycoprotein in nature.

The disease is classified into two types: the primary type or Urbach-Wiethe type and the symptomatic type or

light sensitive form which is designated as erythropoietic protoporphyria.

The light sensitive form usually affects only the exposed skin, mucosal lesions are not observed and there is no hoarseness of voice.

The Urbach-Wiethe type begins from infancy, and occurs equally in both sexes.

The diagnostic criteria are hoarseness since birth, in addition to the mucosal and cutaneous lesions.

The skin manifestations are most striking on the face, forearms and anterior aspects of the 'lower legs, there are elevated yellowish white or brownish papules or hyperkeratotic verrucous lesions, atrophic scars are common and the little pearly papules on the eyelid edges are characteristic. The lips are also affected.

Similar infiltrations may occur in the tongue, the palate, pharyngeal and buccal mucosae with nodules in the larynx and vocal cords.

The mucosa of the vagina , labiae and rectum may be affected also.

Widespread visceral involvement is observed as infiltration of the oesophagus, stomach, pancreas, lung, kidney,

testis and intracranial calcifications with epilepsy.

Diabetes mellitus may be associated. Dental abnormalities frequently coexist with enlargement of parotid glands.

As regards treatment, surgical removal of the infiltrations on the vocal cords may be attempted. No other treatment is known to play any part in influencing the signs and symptoms of the disease.